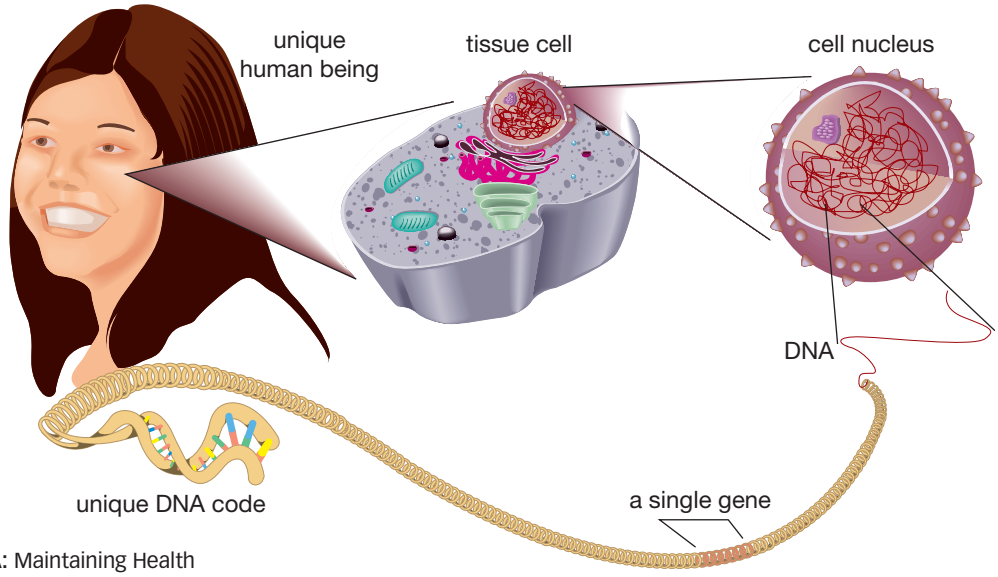
2.1 What Is Genetics?

People come in a wonderful variety of shapes, sizes, and colours. Some aspects of a person's appearance—such as hair style—are changeable and a result of personal grooming choices or current fashion trends. Traits that are not changeable include features such as the natural colour of your skin, your blood type, and whether or not you have dimples in your cheeks. Although it's easy enough to simply say that these traits are inherited, it is considerably more challenging to explain how a person's cells develop in a way that favours one trait over another.

If you were to use a microscope to look closely at any living thing, you would see that the organism is made up of small living units called cells. Some organisms are so tiny that they are made up of just one cell, whereas large and complex organisms are made up of trillions of cells. The cells in your body do all the jobs needed for you to live and remain healthy.

Locating Genetic Information Within the Cell



Chromosomes

In previous science courses you have studied the major parts of a cell. Recall that cells have a region called the nucleus that acts like a command centre to direct cell activity. If you used a very powerful microscope to look deep inside the nucleus of any one of your body's cells, you would find long strands of information called **chromosomes**. Each chromosome can be thought of as a book of instructions: almost like a cookbook full of recipes. A very simple organism, such as a bacterium, has all of its information on a single long strand. More complex organisms, such as people, tend to have several chromosomes. The number of chromosomes in a cell's nucleus depends on the type of species. Humans have 23 pairs of chromosomes or 46 total chromosomes. All the chromosomes in your nuclei are like a library of cookbooks or a complete set of instruction manuals with all the necessary information to run the activities of your cells.

chromosome: a strand of DNA that contains the instructions for making proteins

Chromosomes become X-shaped before cells divide.

Chromosomes are found in pairs. One outcome of this design is that a backup copy of important information is created. However, since one chromosome comes from each parent, there might be slight differences in the instruction subsets. To return to the cookbook analogy, it is like a cookbook with two recipes for chocolate fudge. Despite the fact that the instructions are nearly identical, slight variations in cooking times and temperatures result in food with different textures and consistencies.

Every cell has a complete set of chromosomes even though that particular cell might not need all the information to make a complete organism. The cell only uses the instructions required for its particular needs. This is just like several people who own the same cookbook using different recipes from the same book depending on their individual needs. If you made a cake from a cookbook recipe, you wouldn't have to read through the whole book each time you wanted to make a cake. In the same way, a particular cell from the pancreas only needs to read some of the information. This data could be, for example, a description of how to produce insulin.

Cookbooks and chromosomes differ in an important way. Information in cookbooks is organized by topic with all the recipes for desserts in one chapter or in one specific cookbook in your cookbook library, but the chromosome genes are not organized in a similar way. The instructions for making a hand are not found on one particular chromosome, but the instructions are instead spread among several chromosomes.

Chromosomes are generally long, thin strands that are coiled at regular intervals around protein molecules for protection. Chromosomes are best seen and photographed when the cell is dividing. At this time, each chromosome produces an identical copy of itself. The two copies, which remain attached at one point, shorten by coiling to produce the characteristic X-shape shown in photographs.

Genes

At specific places on each chromosome there are encoded instructions called **genes**. If a chromosome is like a recipe book, then a gene is like a specific recipe that provides the detailed instructions for building certain proteins. In order for instructions to be stored, communicated, and then used to complete a task, a language is necessary. Cookbooks are written in languages—like Korean or German—that people can understand. Genetic instructions are written in a chemical language called **deoxyribonucleic acid** or **DNA** for short. This language is encoded in a molecule. DNA has a distinctive shape, called a double helix, that looks like a twisted ladder or a spiral staircase. The sequence of chemical components in the DNA molecule encodes information. Each rung in the spiral ladder of the DNA molecule can be thought of as an individual letter in the cookbook.

- **gene:** a segment of DNA that carries instructions that result in the production of proteins
- **deoxyribonucleic acid (DNA):** the twisted ladder-shaped molecule that contains the genetic information of cells
- **genetics:** the science of gene function and inheritance

The ability to roll your tongue is controlled by a gene and determined by a certain pattern or sequence along a DNA molecule. People who are unable to roll their tongues do not have the gene instructions that allow the tongue muscles to roll. Even though two people each have the gene instructions for making hair or an eyeball, slightly different recipes create curly hair instead of straight hair or blue eyes rather than brown eyes. The combination of instructions from the genes on your chromosomes determines many of your characteristics. **Genetics** is the scientific study of how genes work to determine characteristics and to resolve how genetic information gets passed from parent to offspring.

Practice

1. **a.** Describe two ways in which chromosomes and cookbooks are similar.
   b. Describe one way that chromosomes and cookbooks are different.
2. How many chromosomes are found in one of the cells in your hand?
3. Why do chromosomes often appear with an X-shape in diagrams and in photos taken through a microscope?
4. Describe what is meant by the term *double helix*.

Human Karyotype

- **karyotype:** an image that organizes the chromosomes of a cell in relation to number, shape, and size
- **centromere:** the region on a replicated chromosome that attaches the two identical copies during cell division

The human body contains 23 pairs of chromosomes, but the chromosomes are not neatly organized in the nucleus. In fact, they are in a jumble that looks like a plate of spaghetti. A **karyotype** is an image of all the chromosomes in one nucleus that have been matched up into their respective pairs and arranged from the largest pair to the smallest pair. A karyotype allows geneticists to better study the chromosomes in a nucleus. Geneticists use three features to identify and match up chromosomes:

- the length of the chromosome (The longest chromosome is numbered as chromosome 1, etc.)
- the pattern of dark bands produced on each chromosome when they are stained
- the position of the chromosome's constricted part, called the **centromere**, which plays a role during cell division

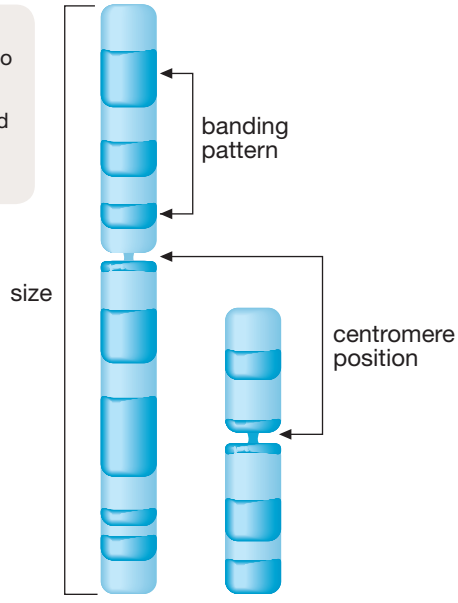


Figure A2.1 illustrates how it is possible for chromosomes to be matched up.

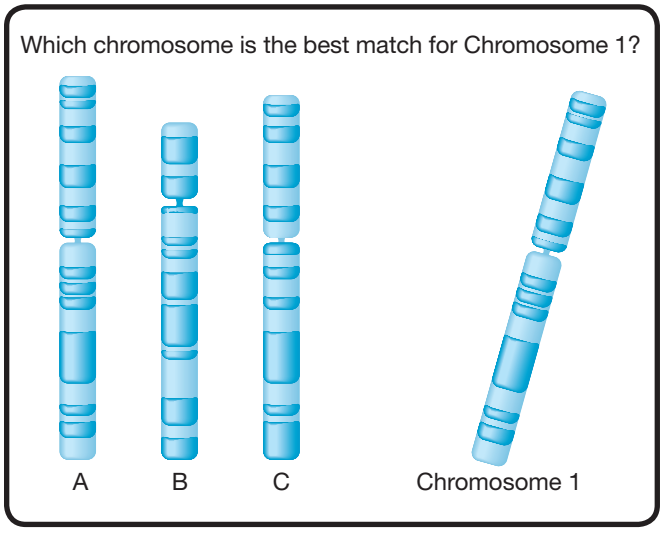
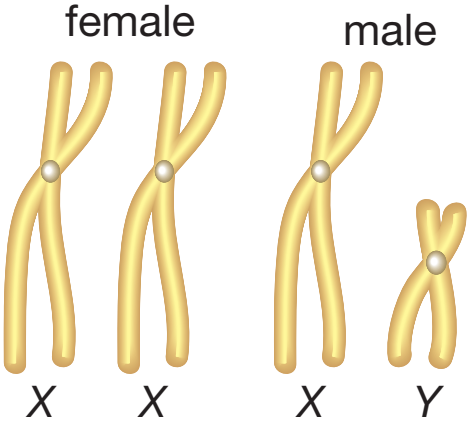


Figure A2.1

An artist's representation of the chromosomes makes the process of chromosome matching look easy—A is clearly the best match for Chromosome 1. However, there are complications when examining photographs of actual chromosomes taken through a microscope—it is challenging to interpret the somewhat grainy photographs of these incredibly tiny objects. Also, the last two chromosomes in the karyotype are called the *sex chromosomes* because they determine the organism's gender.

The pair of chromosomes matched at chromosome 23 determine the sex of the individual.



Note that these chromosomes are shown after replication.

If an individual has two *X* chromosomes, she is a female. If an individual has one *X* chromosome and one *Y* chromosome, he is a male. Since the *Y* chromosome is considerably smaller than the *X* chromosome, this creates an extra challenge for matching chromosomes for a male.

As you'll discover in the next activity, overcoming these challenges is like sorting out a jigsaw puzzle.

Try This Activity

Make a Human Karyotype

To complete this activity you will need a copy of the handouts "Cut and Paste Karyotype Activity" and "Cut and Paste Karyotype Activity—Matched," which are available on the Science 30 Textbook CD.

Science Skills

- ✓ Performing and Recording
- ✓ Analyzing and Interpreting

Purpose

On the handouts you will find the corresponding chromosome that best matches each of the numbered chromosomes to form a human karyotype.

Materials

- "Cut and Paste Karyotype Activity" handout
- "Cut and Paste Karyotype Activity—Matched" handout
- scissors
- glue or transparent adhesive tape

Procedure

step 1: Cut out the 23 unnumbered chromosomes from the handout titled "Cut and Paste Karyotype Activity."

step 2: Place each unnumbered chromosome with a numbered chromosome to produce a matched pair.

step 3: Check your completed karyotype by using the handout named "Cut and Paste Karyotype Activity—Matched."

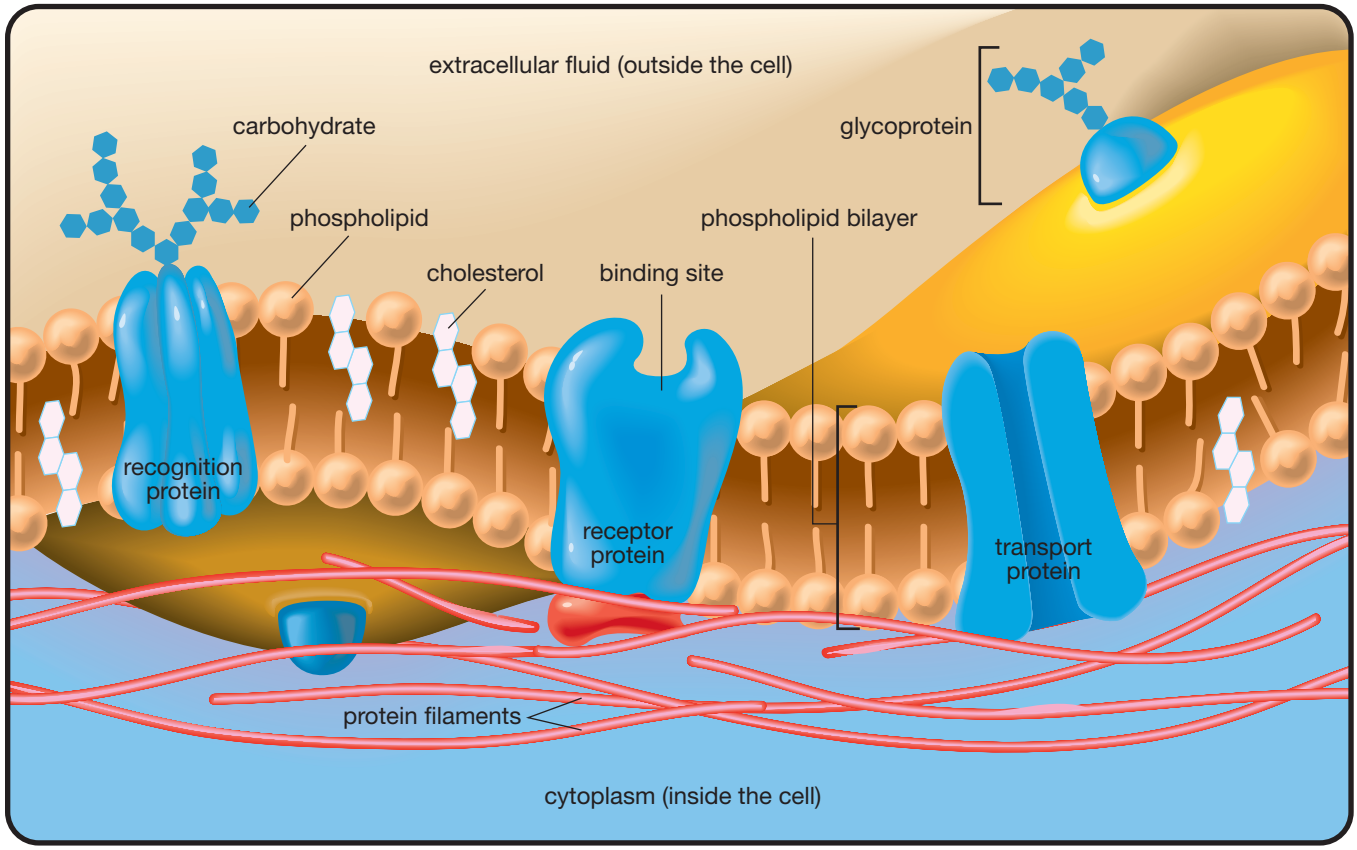
Analysis

1. Why it is beneficial to have two sets of chromosomes? Where does each set of chromosomes come from?
2. **a.** Describe what is different in terms of shape and size in the last pair of chromosomes.
   b. Is this individual a male or a female?
3. Explain how a karyotype of an individual might be useful to scientists.

The Role of Proteins

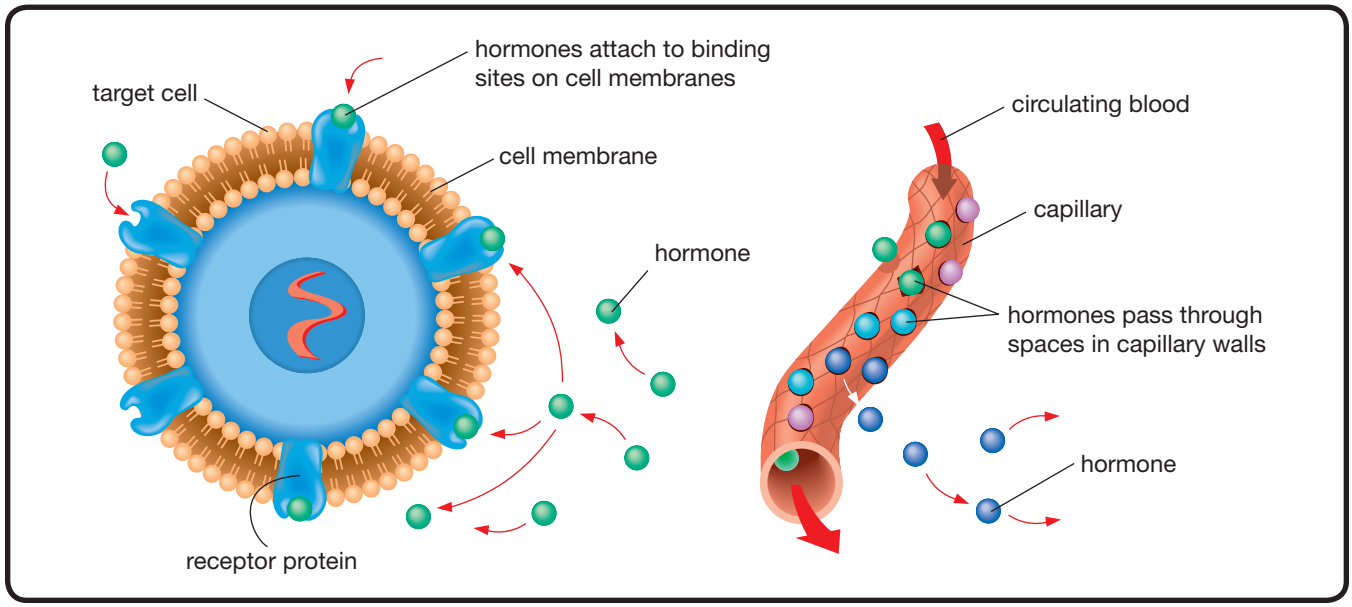
More than 25 000 genes are spread over the 46 chromosomes in the human karyotype. Chromosomes are like cookbooks, and each gene is a recipe for making a specific protein. This means there are more than 25 000 different recipes for proteins. Why is all this vast library of genetic information focused on making proteins? What role do proteins play in the body?

Cell Membrane Proteins



Proteins are molecules that have versatile and important bodily functions. If you took away all the water in the human body, about 50% of the dry mass left behind would be proteins. The human body produces tens of thousands of proteins, each with a unique structure and a specific job. If the body is like an engine, each protein is like a tiny specialized part that completes a specific task to keep the engine running. Hormones are proteins that co-ordinate and regulate the body's activities. This is shown in Figure A2.2.

Figure A2.2



The major types of proteins found in organisms and some of their roles are summarized in the following table.

SUMMARIZING THE ROLES OF PROTEINS

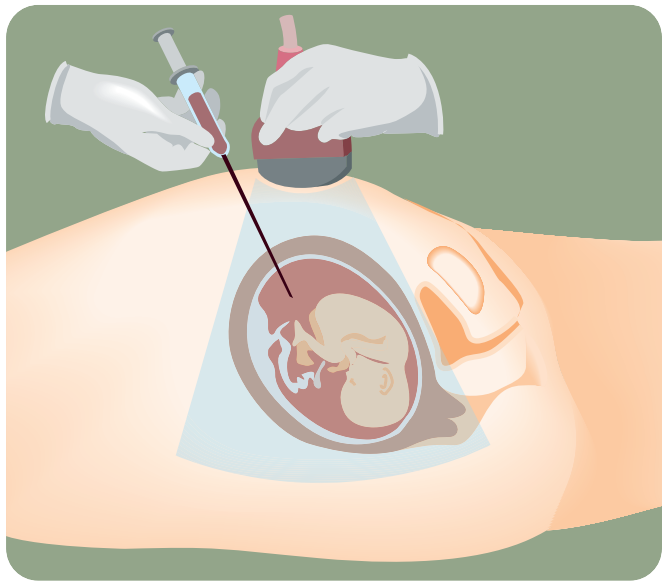
Type of Protein	Role of Protein	Example
enzyme	Enzymes speed up chemical reactions where molecules are broken apart or put together.	Amylase is a digestive enzyme in your saliva that breaks down long starch molecules into shorter, more digestible glucose molecules.
structural	Structural support and frameworks are created to attach to other proteins.	Keratin is a structural protein that makes up your hair and nails. Collagen is a structural protein that provides a framework for skin and internal organs.
transport	Materials are moved within the cell or body.	Cell membrane proteins form channels and pumps in the cell membrane to help needed materials flow into the cell and unwanted materials flow out of the cell.
hormone	Hormones act as signals to co-ordinate and regulate activities in the body.	Insulin is a hormonal protein that regulates blood sugar. Insulin is produced in the pancreas and moves in the bloodstream to other organs to influence their use of glucose.
contractile	Contractile proteins change shape and can create larger movements when they work together.	Actin and myosin are proteins that band together to allow muscles to contract.
defensive	Defensive proteins protect the body against disease.	Antibodies are proteins that act in the body by attaching to disease-causing pathogens and foreign material.
energy	Energy proteins serve as a source of chemical potential energy that can be released by its decomposition.	Casein is an energy protein found in milk.

Practice

5. Identify examples of the kinds of molecules genes are designed to produce.
6. Describe the function of proteins in cell membranes.

Amniocentesis

An **amniocentesis** is a prenatal test done to look at the karyotype of an unborn child. During the test, a small amount of amniotic fluid is drawn out with a large needle from the area around the fetus. An image produced with ultrasound helps to direct the path of the needle. Some of the developing baby's cells are floating in the amniotic fluid and these cells are examined for genetic abnormalities, such as the presence of additional chromosomes, as is the case in Down syndrome. Amniocentesis is not routinely offered to expectant mothers because the procedure can slightly increase the chance of miscarriage, and it is only performed in pregnancies where there is a high risk of genetic diseases or deformities. Factors that can increase the risk of genetic diseases or deformities include the mother's age (above 40) or a history of severe diseases known to have a genetic origin.



amniocentesis: a prenatal test done to look at the karyotype of an unborn child

Making More Chromosomes

When a cell divides, it must provide genetic information to each of the new cells that form from the cell division. This means that exact copies must be made of the long strands of DNA within each of the chromosomes. Depending upon the type of cell, there are two basic ways in which this process can occur. These methods are mitosis and meiosis.

Mitosis

In order to grow and to replace cells that are dead or damaged, your body must constantly make new cells. Skin cells, for example, need to be frequently replaced by new cells. A body cell, or an **autosomal cell**, divides by growing large and making extra copies of all its parts and then splitting into two. An autosomal cell is shown at the top of Figure A2.3. Instead of showing 23 pairs of chromosomes, the simplified illustration shows only one pair. Note that one chromosome from the pair is inherited from the father and the other from the mother. Since each chromosome within the pair carries genes for the same characteristics at the same chromosome location, the pair of chromosomes are called **homologous chromosomes**.

To ensure that new cells have the necessary genetic information, autosomal cells must make a copy of their chromosomes before dividing. It is said that the DNA **replicates**. The replicated chromosomes attach at the centromere to form a distinctive X-shape. If each of these chromosomes is thought of as a recipe book, the process of DNA replication increases the number of copies of each book from two to four.

After being replicated, the chromosomes move to line up along the cell's middle or equator. The duplicate strands of the chromosomes are then pulled apart. The cell membrane pinches in to split the cell into two new cells with two sets of chromosomes called **daughter cells**. The process of cell division in autosomal cells is called **mitosis**. Note that with two copies of each chromosome, daughter cells are identical to the original autosomal cell that began the whole process. Biologists refer to the original cell and the daughter cells as **diploid cells** because each of these cells has two copies of each chromosome type.

Some organisms are able to produce a new organism by mitosis. This is called **asexual reproduction**. Offspring produced asexually are genetically identical to their parents. Bacteria reproduce asexually by simply splitting in two. A strawberry plant or a spider plant often makes a small copy of itself that breaks off or shoots off on a runner and grows into a new plant. Growing this small copy is called *budding*. The ability of plants to reproduce asexually allows people to use plant cuttings to grow into a whole plant. This plant is a genetic copy of its parent plant.

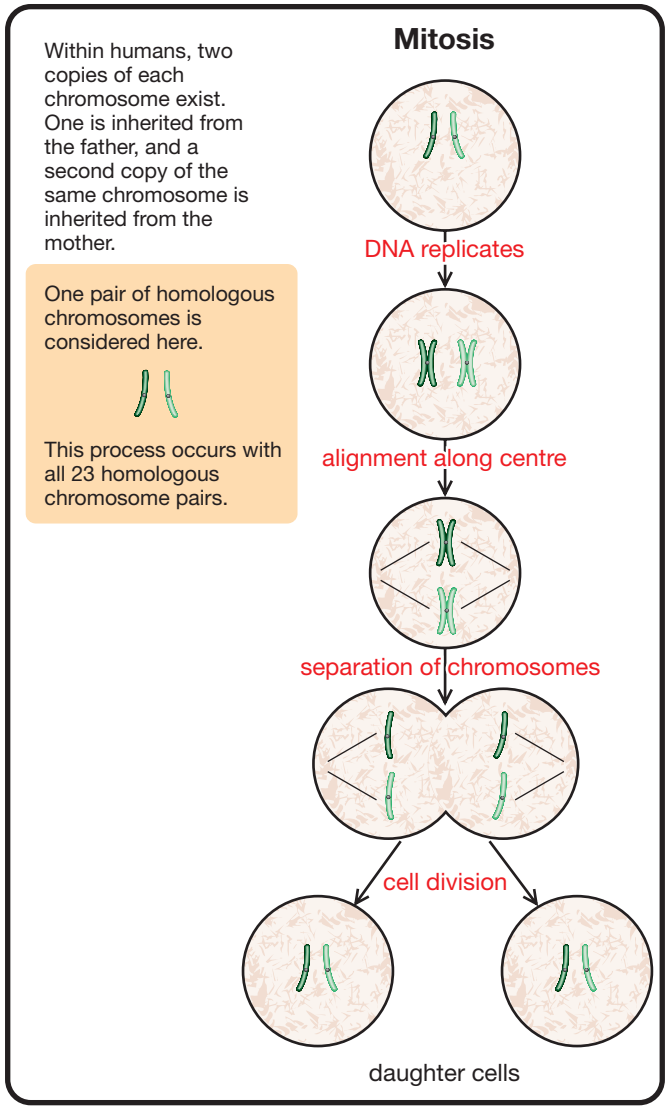


Figure A2.3

- **autosomal cell:** a cell of the body not involved in sexual reproduction
- **homologous chromosomes:** a pair of chromosomes that would be matched during karyotyping because they have the same length, centromere position, and staining pattern
- **replicate:** to produce an exact copy of a DNA strand
- **daughter cells:** the two identical cells produced during mitosis
- **mitosis:** the division of an autosomal cell into two identical daughter cells
- **diploid cells:** cells with pairs of homologous chromosomes
- **asexual reproduction:** the production of genetically identical offspring from one individual

Meiosis

Most organisms do not reproduce asexually. Instead, they produce special reproductive cells called sex cells or **gametes**. These cells combine to make new and unique offspring. In animals the gametes are called sperm and egg, and in plants they are called pollen and egg. The process of producing gametes, called **meiosis**, begins in the same way as mitosis. Cells in the reproductive organs start with chromosomes in homologous pairs. Again, instead of showing 23 pairs, only one pair is noted in Figure A2.4. The DNA replicates and the replicated chromosomes form the characteristic X-shape.

Meiosis begins to differ from mitosis in the steps that follow. The first difference that occurs is that homologous chromosomes pair up and exchange parts. Some DNA segments of the chromosome from one parent are exchanged for corresponding DNA segments on the other parent's chromosome. This process of exchanging genetic material during meiosis is called **crossing over**. Since the pattern of DNA has been altered, crossing over creates slight genetic differences in the chromosomes.

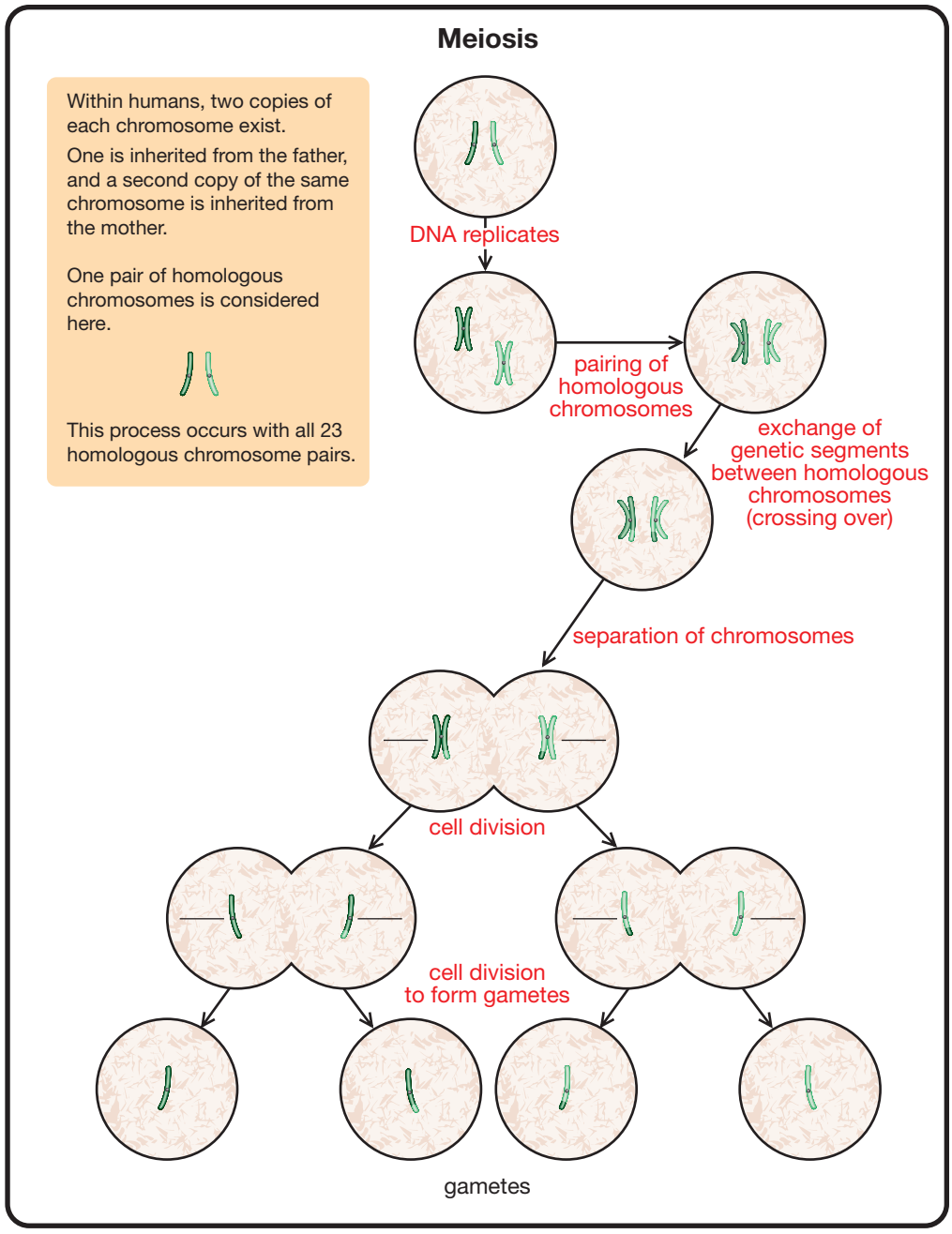


Figure A2.4

After crossing over, chromosomes align along the centre of the cell so that the homologous chromosomes are side by side. The homologous chromosomes separate and two daughter cells are produced. Next, a second cell division occurs, resulting in four cells that are called gametes.

Compare the number of chromosomes in the gametes with the number in the original cell at the top of Figure A2.4. Since there was no DNA replication prior to this second cell division, the four gametes each carry only one set of chromosomes rather than two sets in autosomal cells. The gametes are referred to as **haploid cells**, as they only have half the number of chromosomes as do autosomal cells.

- **gamete:** a sex cell, such as a sperm and an egg, produced during meiosis with only one copy of each chromosome type
- **meiosis:** a two-stage form of cell division that produces gametes with only half of the number of chromosomes as the original cell
- **crossing over:** the exchange of corresponding segments of DNA between maternal and paternal chromosomes during meiosis
- **haploid cell:** a cell that has only one member from each pair of homologous chromosomes

In people there are 23 different types of chromosomes. Biologists use the shorthand $n = 23$ to communicate this idea. Each gamete, either a sperm cell or an egg cell, has only one copy of each of these chromosomes. The notation for these cells is 1n, meaning one copy of each type of chromosome.

Each of the other body cells—the autosomal cells—has two copies of each type of chromosome. These cells are described as 2n cells, meaning two copies of each chromosome. The following memory device can help you remember these ideas.

Mitosis is remembered as "mi two sis."	**Meiosis is remembered as "mei one sis."**
• produces diploid cells (**2n**), with **t**wo copies of each chromosome	• produces haploid cells (**1n**), with **o**ne copy of each chromosome

Practice

7. Obtain a copy of the handout "Mitosis" from the Science 30 Textbook CD.

- **a.** Add the missing labels to "Mitosis."
- **b.** Add the labels "1n" and "2n" to describe the original cell and the daughter cells.

8. Obtain a copy of the handout called "Meiosis" from the Science 30 Textbook CD.

- **a.** Add the missing labels to this diagram.
- **b.** Add the labels "1n" and "2n" to describe the original cell and the gametes.

Utilizing Technology

Comparing Mitosis and Meiosis

Background Information

Now that you have seen the details of mitosis and meiosis, it's important to be able to keep the big picture in mind and not to lose sight of the main ideas. This activity provides an opportunity to reinforce essential concepts about mitosis and meiosis.

Science Skills

✓ Performing and Recording

Purpose

You will use an applet titled "What Is Mitosis/Meiosis?" to reinforce the essential concepts that are necessary to understand mitosis and meiosis.

Procedure and Observations

step 1: Locate the applet "What Is Mitosis/Meiosis?" from the Science 30 Textbook CD.

step 2: Review the list of analysis questions. Then watch the applet.

step 3: Watch the applet again, only this time use the natural pauses between screens to record your answers.

Analysis

1. A human being begins as one cell and then grows into a body of a hundred trillion cells.

- **a.** Define the term *diploid cell*.
- **b.** Describe the arrangement of chromosomes in human body cells.
- **c.** Explain why it is necessary, before it divides, for a body cell to make a copy of each chromosome.
- **d.** Identify the process that describes how one cell becomes a body of trillions of cells.

2. Not all cells in the human body are diploid cells.

- **a.** Define the term *haploid cell*.
- **b.** Describe in general terms the process that produces haploid cells.
- **c.** Identify the name of the process that you described in question 2.b.
- **d.** Explain the purpose of haploid cells.
- **e.** Explain what happens when two haploid cells combine.

3. Describe the essential difference between mitosis and meiosis.

Fertilization

When chromosomes in a male gamete join up with chromosomes in a female gamete during fertilization, the fused cell contains the two sets of chromosomes found in an autosomal cell. With two sets of chromosomes, the fertilized egg is able to grow and develop through the process of mitosis.

The advantage of sexual reproduction is that diverse offspring are produced. The genetic combination of chromosomes from each parent results in offspring with the possibility of different traits. As shown in Figure A2.5, even with a simplified model using only one pair of chromosomes, there are 16 possible outcomes. If the full complement of 23 pairs of chromosomes are used, there will be more than 70 trillion possible ways for gametes from a mother and a father to join and create a new human being. The number of possibilities would be even greater if crossing over was considered. Given that this number is more than all the people who have ever lived on planet Earth, unless you are an identical twin, there has never been anyone with your exact DNA. In other words, you are a unique creation.

Fertilization

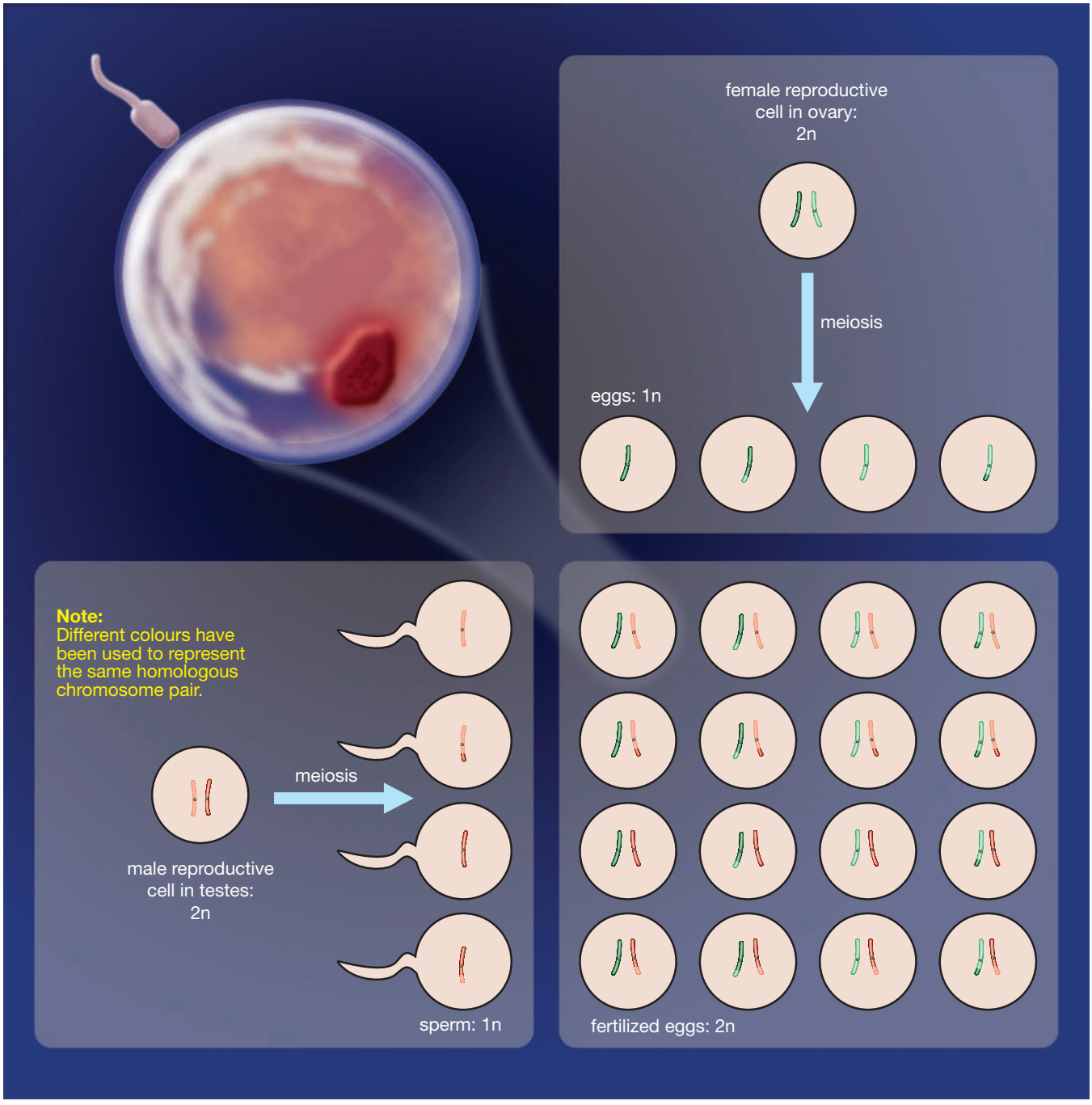


Figure A2.5: This simplified model shows the outcomes from 1 pair of chromosomes instead of 23 pairs.

Sexual reproduction is clearly a valuable mechanism for increasing the variety among organisms in a population. Note the supporting role that crossing over plays to increase variety in the gametes produced by meiosis. The exchange of genetic material during crossing over further increases the genetic diversity of the offspring, and this is part of the reason why two siblings from the same parents can look quite different. Genetic diversity is important because a genetically diverse population is less susceptible to disease.

Utilizing Technology

Determining Numbers of Unique Offspring

Purpose

You will use a spreadsheet to verify the following statement:

> " . . . there would be more than 70 trillion possible ways that gametes from a mother and a father can join to create a new human being."

Science Skills

✓ Analyzing and Interpreting

Procedure

You may complete this activity by using a spreadsheet or a graphing calculator. Use the following template as a guide. Continue the patterns shown until the Number of Pairs of Chromosomes column reaches 23 pairs. At this point, the table is complete.

Number of Pairs of Chromosomes	Number of Possible Gametes Produced by One Parent (excluding crossing over)	Number of Possible Unique Combinations of Offspring Produced by Two Parents
n	2^n	$(2^n) \times (2^n)$
1	$2^1 = 2$	$(2^1) \times (2^1) = 2 \times 2 = 4$
2	$2^2 = 4$	$(2^2) \times (2^2) = 4 \times 4 = 16$
3	$2^3 = 8$	$(2^3) \times (2^3) = 8 \times 8 = 64$
4	$2^4 = 16$	
5		

Analysis

If the number of gametes produced by each parent is 2^n, explain why the number of offspring produced by two parents is $(2^n) \times (2^n)$.

Selective Breeding

Long before chromosomes and genes were discovered, people recognized that offspring had a similar appearance to their parents and hypothesized that characteristics could be inherited. However, people were not able to explain how these characteristics were passed on from parents to offspring. They lacked necessary tools—such as microscopes powerful enough to see cells and their parts—to understand mechanisms for the inheritance of traits or the genes responsible for them.

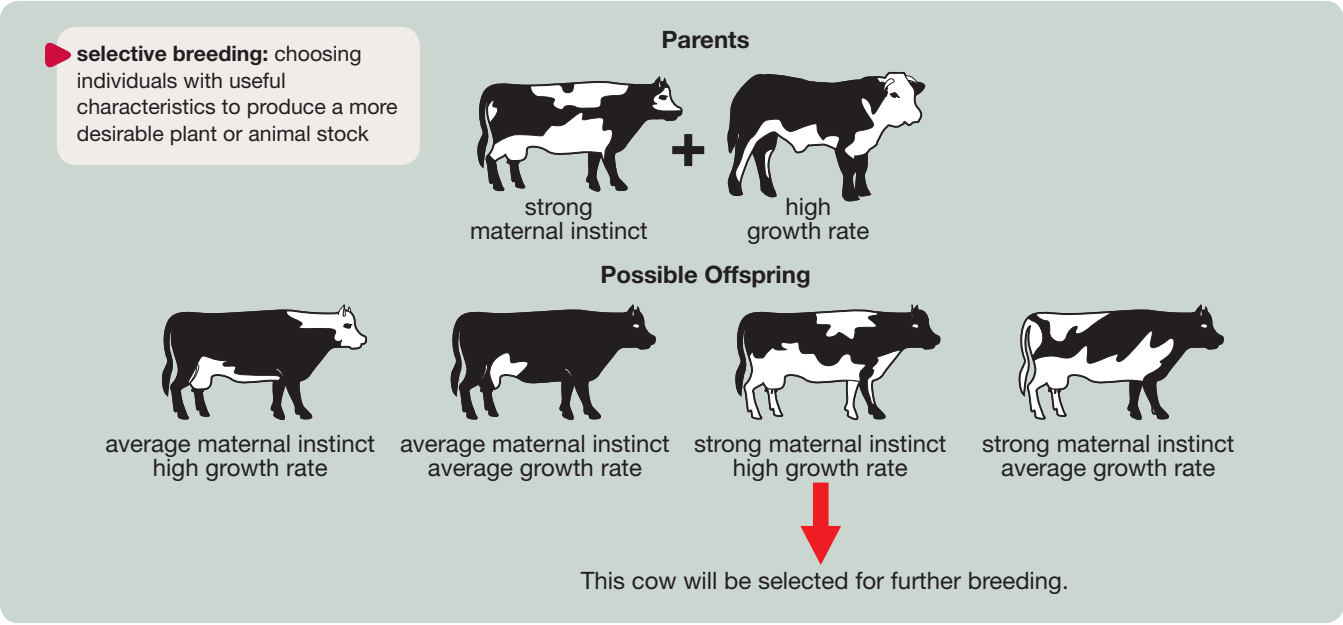


Figure A2.6: Raising cattle for beef production involves selective breeding. A strong maternal instinct in females means good nurturing and feeding of calves. Orphaned calves often grow poorly or they may die. A high growth rate demonstrates that the animal's metabolism is able to convert ingested food into tissue (meat).

Ancient peoples who lived in aristocratic societies believed that the monarchs passed on their noble or royal blood to their children. These people also realized that they could select individuals with desirable characteristics from their domestic crops and livestock. These crops and livestock could then be bred to produce offspring possessing desirable traits. Farmers could also prevent individuals with undesirable characteristics from breeding. The practice of **selective breeding** allowed farmers to create stocks that met their particular needs.

Figure A2.7: Corn husks were used by Iroquois to make masks or false faces that had special spiritual significance in healing ceremonies.

First Peoples of North America and Central America employed selective breeding to grow domesticated crops—such as maize (corn)—for thousands of years before Europeans arrived in the area. In 1612, French explorers around the Great Lakes recorded that Haudenosaunee (Iroquois) people made popcorn by putting maize kernels in pottery full of heated sand. It is believed that the maize First Nations people grew is a result of centuries of selective breeding of teosinite, a wild grass. The corn and popcorn that you eat today is descended from the breeds developed by First Nations people.

Practice

Use the following information to answer question 9.

In previous courses you studied the theory of natural selection. This theory states that evolution takes place because more organisms are produced than can survive and that only the organisms best suited to their environment survive to reproduce and, in turn, pass on their advantageous traits to their offspring. According to this theory, the environment determines what organisms will be successful and will therefore have their traits determine the characteristics seen in future generations.

9. a. Describe how selective breeding differs from the process of natural selection.

b. Describe how selective breeding is similar to the process of natural selection.

10. Genetic engineering can be defined as the manipulation of genes in organisms to produce desirable characteristics and to eliminate undesirable characteristics.

a. Explain how selective breeding is a form of genetic engineering.

b. Identify the first people to practise genetic engineering in Canada.

Early Ideas of Inheritance

The development of simple microscopes allowed for the discovery of cells, such as sperm and eggs; but these instruments were not powerful enough to observe chromosomes. During the seventeenth and eighteenth centuries, the theory of preformation was popular. Preformation is the idea that all body parts are already formed at the beginning of development and simply grow from a very tiny full-formed body to a larger body. Prominent scientists including Anton van Leeuwenhoek and Marcello Malpighi, the person who discovered capillaries, were supporters of the preformationist theory. However, there was some debate about whether the pre-formed body was found in the sperm or the egg.

Figure A2.8: Charles Darwin

In the nineteenth century, Charles Darwin used his studies of the natural world to develop and publish his theory of evolution. His theory stated that organisms best suited to their environment survived to pass on their characteristics to their offspring, but Darwin was not able to explain how characteristics are passed on.

Discovering Genes

In the 1860s, Gregor Mendel was the first person to undertake detailed and systematic studies into the inheritance of characteristics. Mendel was a monk who became interested in the breeding of pea plants that grew in the monastery where he lived. Little did he know that he would become known as "the father of genetics."

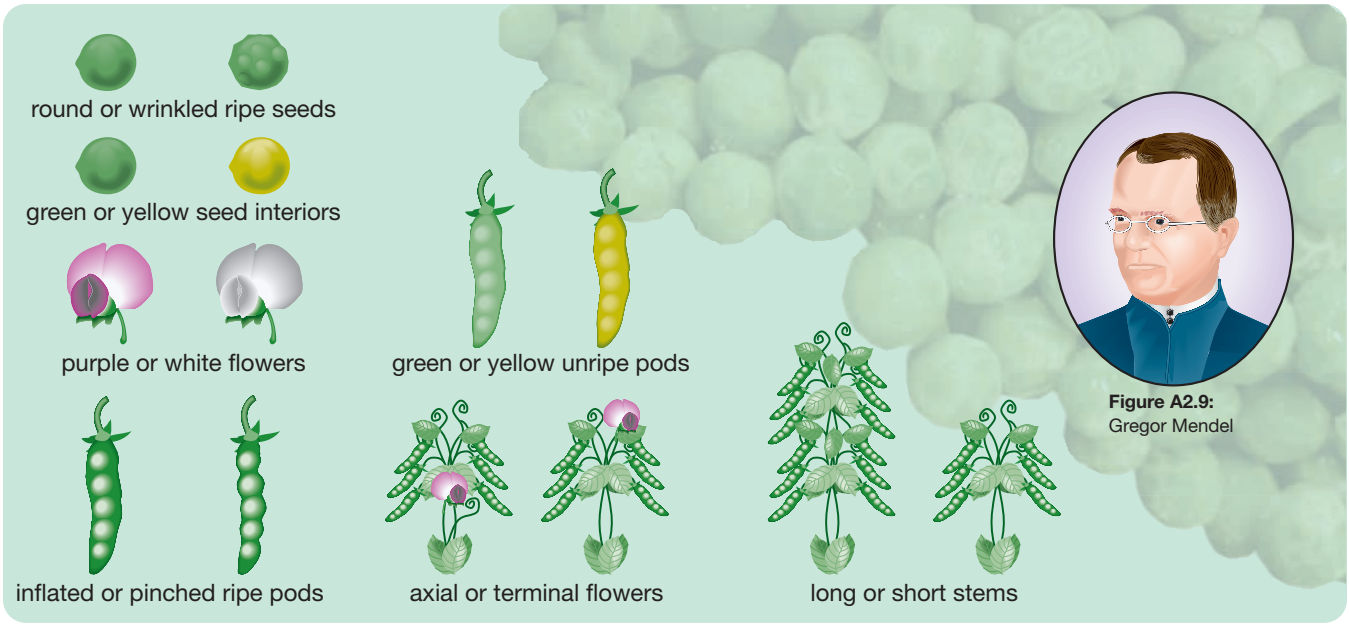


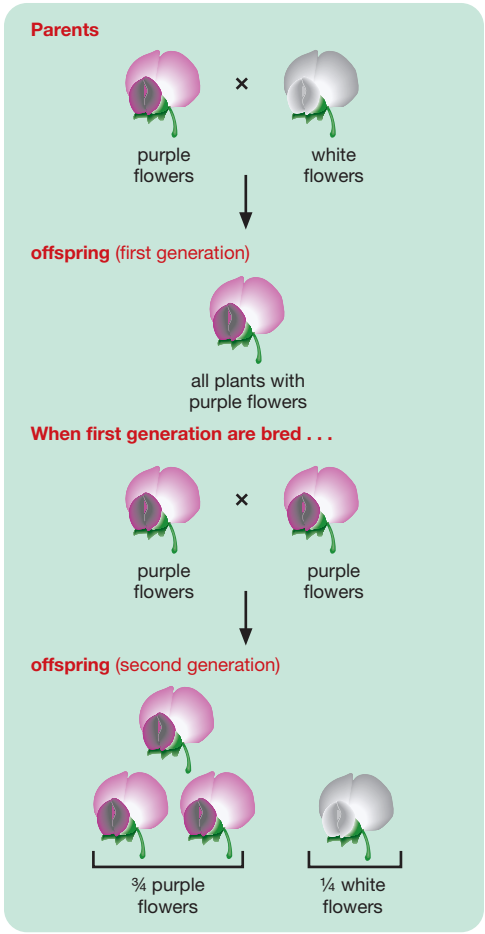
Figure A2.9: Gregor Mendel

Mendel noticed that certain inherited characteristics were not blended. For example, a pea plant with white flowers when bred with a pea plant with purple flowers did not produce offspring with blended characteristics. Instead, the offspring had either purple flowers or white flowers.

Common garden pea plants were good test subjects for his research because not only are they easy to grow in large numbers, but their reproduction can be manipulated by transferring pollen from one plant to another in a process called **cross-pollinating**. Mendel discovered that several traits in pea plants were easy to recognize and occurred in only one of two distinctive forms—purple flowers or white flowers, round seeds or wrinkled seeds, yellow seeds or green seeds, and other forms.

- **cross-pollinate:** transferring pollen between genetically different plants
- **self-pollinate:** transferring pollen from one plant to the female part of the same plant or to another plant with the same genetic makeup

Mendel observed that when he cross-pollinated a white flowering plant with a purple flowering plant, the offspring from this cross all had purple flowers. The white-flowered trait had disappeared in the generation of pea plants produced by this cross. When he bred individuals from this first generation, or allowed them to **self-pollinate**, he was surprised to see that the white-flowered trait returned in about a quarter of the second-generation's population. Experiments on other pea traits provided the same results. One trait completely disappeared in the first generation and then re-appeared in about a quarter of the second-generation plants.



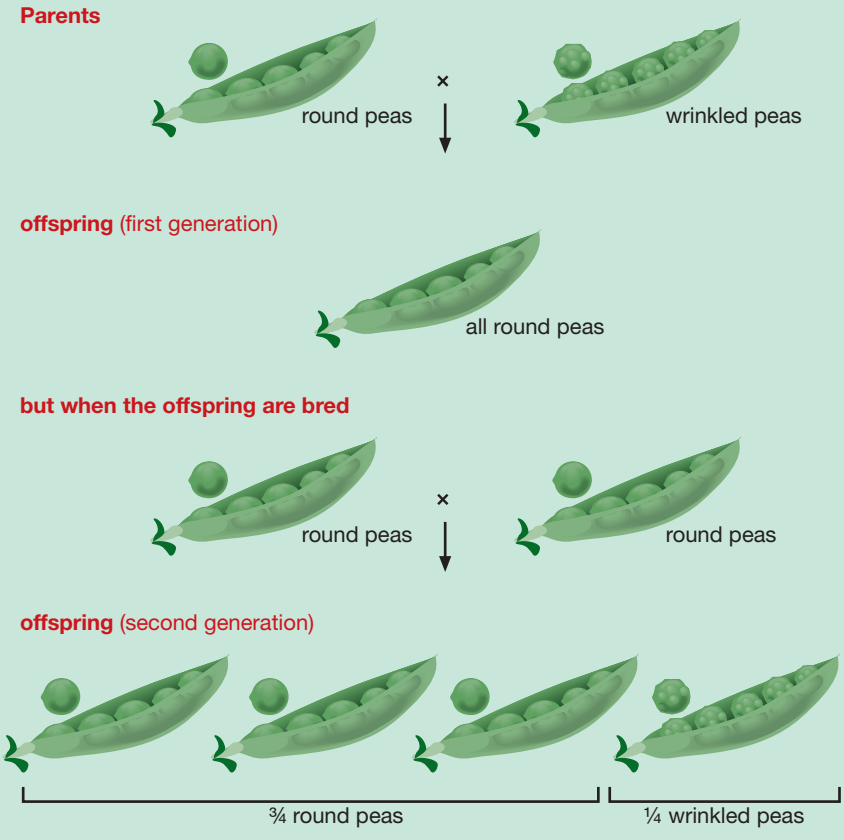
Mendel's Conclusions from His Pea Plant Experiments

(1) The inheritance of traits must be determined by factors (now called genes).

(2) Individuals randomly inherit one factor (copy of a gene) from each parent.

(3) Factors or genes are independently passed to the offspring. This means that a pea plant's inheritance of the gene that makes purple flowers does not affect the plant's inheritance of other traits, such as seed colour.

(4) Some genes are more powerful than others; so a trait may not appear in an individual because the more powerful gene masks it (e.g., purple flowers over white flowers), but it can still be passed onto the individual's offspring.



Even though Mendel was studying pea plants, he actually described the basic principles of genetic inheritance for all complex life forms, including people. Mendel published his results in 1866. Sadly, in his lifetime Mendel's results were not understood and the importance of his work was unrecognized. Sixteen years after Mendel's death, chromosomes were discovered and it was suggested that Mendel's factors might be carried on the chromosomes. After his death, Mendel's experiments were repeated by other scientists and his ideas and important contributions to the understanding of how traits are passed from parents to offspring were acknowledged.

2.1 Summary

A chromosome is a long strand of information made up of a double helix molecule called deoxyribonucleic acid or DNA. Chromosomes are found in pairs in the nuclei of autosomal cells. Genes are specific regions along the chromosomes and have the instructions for making proteins. Genetics is the study of how genes work to determine characteristics and how they get passed on.

A karyotype is an image of all the chromosomes paired up and organized for study by geneticists. The human karyotype is composed of 46 chromosomes, 22 pairs of autosomes, and 1 pair of sex chromosomes. The sex chromosomes determine the gender of an individual. Two *X* chromosomes produce a female, and one *X* chromosome combined with one *Y* chromosome produces a male.

Body cells, or autosomal cells, divide for an organism to grow or replace cells through the process of mitosis. During mitosis, two identical daughter cells are produced with identical sets of chromosomes. Some organisms can reproduce asexually through mitosis alone.

Sex cells, or gametes, are produced for sexual reproduction through the process of meiosis. During meiosis, four gametes are produced—each has half the number of chromosomes of an autosomal cell. The combination of male and female gametes creates a fertilized egg with the required two complete sets of chromosomes.

Selective breeding is a technology that has allowed people to develop more useful breeds of domesticated plants and animals to suit human needs.

Mendel's study of pea plants led to the development of a theory proposed where traits were inherited by factors (later called genes) and in which offspring randomly received a copy of a gene for each trait from its parents. Mendel also stated that possessing a gene for one trait did not affect the genes for other traits and that some genes were able to mask the other forms of an inherited trait.

2.1 Questions

Knowledge

1. Match the following terms with the analogy that best describes each term.

- DNA
- gene
- protein
- chromosome
- karyotype

a. a cookbook with several recipes in it

b. an entire library of cookbooks neatly arranged in order from the largest book to the smallest book

c. the cake produced by following recipe instructions

d. the letters and words in a recipe

e. the instructions for making a cake

2. How many chromosomes are in a normal human autosomal cell? How many chromosomes are in a normal human gamete?

3. Determine the gender of an individual who has two *X* chromosomes in each autosomal cell instead of an *X* and a *Y* chromosome.

4. Dogs have 78 chromosomes, cats have 34 chromosomes, and goldfish have 94 chromosomes. Explain why the usual number of chromosomes in autosomal cells for any species is always an even number.

5. Describe what would happen if the process of meiosis did not occur and two cells with two sets of chromosomes combined to produce a new child. Why is meiosis necessary?

6. Identify the term Mendel used instead of *gene*.

7. List some reasons why pea plants are so well suited for genetic studies.

Applying Concepts

8. Obtain a copy of the handout "Comparing Mitosis and Meiosis," from the Science 30 Textbook CD.

a. Without looking at information from this lesson, attempt to add the missing labels to this diagram.

b. Biologists use the letter *n* to represent the number of different chromosomes in a cell. Label the cells at the top and bottom of the handout either 1n or 2n.

c. Use the information in Lesson 2.1 to check and correct your answers to 8.a.

9. Sexual reproduction creates beneficial genetic diversity. List some possible advantages of asexual reproduction.

10. Explain the steps you would take in the process of selective breeding to create a fast-running breed of dog.

11. Propose some reasons why van Leeuwenhoek and other scientists of his time believed in the preformation theory.